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<!--StartFragment-->RESULT 7
MYPR_HUMAN
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AC   P60201; P04400; P06905; Q502Y1;
DT   01-JAN-1988, integrated into UniProtKB/Swiss-Prot.
DT   23-JAN-2007, sequence version 2.
DT   08-APR-2008, entry version 56.
DE   Myelin proteolipid protein (PLP) (Lipophilin).
GN   Name=PLP1; Synonyms=PLP;
OS   Homo sapiens (Human).
OC   Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi;
OC   Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini;
OC   Catarrhini; Hominidae; Homo.
OX   NCBI_TaxID=9606;
RN   [1]
RP   NUCLEOTIDE SEQUENCE [GENOMIC DNA].
RX   MEDLINE=87092337; PubMed=3467339;
RA   Diehl H.-J., Schaich M., Budzinski R.-M., Stoffel W.;
RT   "Individual exons encode the integral membrane domains of human myelin
RT   proteolipid protein.";
RL   Proc. Natl. Acad. Sci. U.S.A. 83:9807-9811(1986).
RN   [2]
RP   NUCLEOTIDE SEQUENCE [MRNA] (ISOFORM DM-20).
RX   MEDLINE=87298492; PubMed=2441695; DOI=10.1016/0006-291X(87)90580-8;
RA   Simons R., Alon N., Riordan J.R.;
RT   "Human myelin DM-20 proteolipid protein deletion defined by cDNA
RT   sequence.";
RL   Biochem. Biophys. Res. Commun. 146:666-671(1987).
RN   [3]
RP   NUCLEOTIDE SEQUENCE [MRNA], AND VARIANT PMD ARG-163.
RX   MEDLINE=90046751; PubMed=2479017;
RA   Hudson L.D., Puckett C., Berndt J., Chan J., Gencic S.;
RT   "Mutation of the proteolipid protein gene PLP in a human X chromosome-
RT   linked myelin disorder.";
RL   Proc. Natl. Acad. Sci. U.S.A. 86:8128-8131(1989).
RN   [4]
RP   NUCLEOTIDE SEQUENCE [LARGE SCALE GENOMIC DNA].
RX   PubMed=15772651; DOI=10.1038/nature03440;
RA   Ross M.T., Grafham D.V., Coffey A.J., Scherer S., McLay K., Muzny D.,
RA   Platzer M., Howell G.R., Burrows C., Bird C.P., Frankish A.,
RA   Lovell F.L., Howe K.L., Ashurst J.L., Fulton R.S., Sudbrak R., Wen G.,
RA   Jones M.C., Hurles M.E., Andrews T.D., Scott C.E., Searle S.,
RA   Ramser J., Whittaker A., Deadman R., Carter N.P., Hunt S.E., Chen R.,
RA   Cree A., Gunaratne P., Havlak P., Hodgson A., Metzker M.L.,
RA   Richards S., Scott G., Steffen D., Sodergren E., Wheeler D.A.,
RA   Worley K.C., Ainscough R., Ambrose K.D., Ansari-Lari M.A., Aradhya S.,
RA   Ashwell R.I., Babbage A.K., Bagguley C.L., Ballabio A., Banerjee R.,
RA   Barker G.E., Barlow K.F., Barrett I.P., Bates K.N., Beare D.M.,
RA   Beasley H., Beasley O., Beck A., Bethel G., Blechschmidt K., Brady N.,
RA   Bray-Allen S., Bridgeman A.M., Brown A.J., Brown M.J., Bonnin D.,
RA   Bruford E.A., Buhay C., Burch P., Burford D., Burgess J., Burrill W.,
RA   Burton J., Bye J.M., Carder C., Carrel L., Chako J., Chapman J.C.,
RA   Chavez D., Chen E., Chen G., Chen Y., Chen Z., Chinault C.,
RA   Ciccodicola A., Clark S.Y., Clarke G., Clee C.M., Clegg S.,
RA   Clerc-Blankenburg K., Clifford K., Cobley V., Cole C.G., Conquer J.S.,
RA   Corby N., Connor R.E., David R., Davies J., Davis C., Davis J.,
RA   Delgado O., Deshazo D., Dhami P., Ding Y., Dinh H., Dodsworth S.,
RA   Draper H., Dugan-Rocha S., Dunham A., Dunn M., Durbin K.J., Dutta I.,
RA   Eades T., Ellwood M., Emery-Cohen A., Errington H., Evans K.L.,
RA   Faulkner L., Francis F., Frankland J., Fraser A.E., Galgoczy P.,
RA   Gilbert J., Gill R., Gloeckner G., Gregory S.G., Gribble S.,
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RA   Griffiths C., Grocock R., Gu Y., Gwilliam R., Hamilton C., Hart E.A.,
RA   Hawes A., Heath P.D., Heitmann K., Hennig S., Hernandez J.,
RA   Hinzmann B., Ho S., Hoffs M., Howden P.J., Huckle E.J., Hume J.,
RA   Hunt P.J., Hunt A.R., Isherwood J., Jacob L., Johnson D., Jones S.,
RA   de Jong P.J., Joseph S.S., Keenan S., Kelly S., Kershaw J.K., Khan Z.,
RA   Kioschis P., Klages S., Knights A.J., Kosiura A., Kovar-Smith C.,
RA   Laird G.K., Langford C., Lawlor S., Leversha M., Lewis L., Liu W.,
RA   Lloyd C., Lloyd D.M., Loulseged H., Loveland J.E., Lovell J.D.,
RA   Lozado R., Lu J., Lyne R., Ma J., Maheshwari M., Matthews L.H.,
RA   McDowall J., McLaren S., McMurray A., Meidl P., Meitinger T.,
RA   Milne S., Miner G., Mistry S.L., Morgan M., Morris S., Mueller I.,
RA   Mullikin J.C., Nguyen N., Nordsiek G., Nyakatura G., O'dell C.N.,
RA   Okwuonu G., Palmer S., Pandian R., Parker D., Parrish J.,
RA   Pasternak S., Patel D., Pearce A.V., Pearson D.M., Pelan S.E.,
RA   Perez L., Porter K.M., Ramsey Y., Reichwald K., Rhodes S.,
RA   Ridler K.A., Schlessinger D., Schueler M.G., Sehra H.K.,
RA   Shaw-Smith C., Shen H., Sheridan E.M., Shownkeen R., Skuce C.D.,
RA   Smith M.L., Sotheran E.C., Steingruber H.E., Steward C.A., Storey R.,
RA   Swann R.M., Swarbreck D., Tabor P.E., Taudien S., Taylor T.,
RA   Teague B., Thomas K., Thorpe A., Timms K., Tracey A., Trevanion S.,
RA   Tromans A.C., d'Urso M., Verduzco D., Villasana D., Waldron L.,
RA   Wall M., Wang Q., Warren J., Warry G.L., Wei X., West A.,
RA   Whitehead S.L., Whiteley M.N., Wilkinson J.E., Willey D.L.,
RA   Williams G., Williams L., Williamson A., Williamson H., Wilming L.,
RA   Woodmansey R.L., Wray P.W., Yen J., Zhang J., Zhou J., Zoghbi H.,
RA   Zorilla S., Buck D., Reinhardt R., Poustka A., Rosenthal A.,
RA   Lehrach H., Meindl A., Minx P.J., Hillier L.W., Willard H.F.,
RA   Wilson R.K., Waterston R.H., Rice C.M., Vaudin M., Coulson A.,
RA   Nelson D.L., Weinstock G., Sulston J.E., Durbin R.M., Hubbard T.,
RA   Gibbs R.A., Beck S., Rogers J., Bentley D.R.;
RT   "The DNA sequence of the human X chromosome.";
RL   Nature 434:325-337(2005).
RN   [5]
RP   NUCLEOTIDE SEQUENCE [LARGE SCALE MRNA] (ISOFORMS 1 AND DM-20).
RC   TISSUE=Spinal cord, and Uterus;
RX   PubMed=15489334; DOI=10.1101/gr.2596504;
RG   The MGC Project Team;
RT   "The status, quality, and expansion of the NIH full-length cDNA
RT   project: the Mammalian Gene Collection (MGC).";
RL   Genome Res. 14:2121-2127(2004).
RN   [6]
RP   PROTEIN SEQUENCE OF 2-277.
RX   MEDLINE=86000127; PubMed=4041237;
RA   Stoffel W., Giersiefen H., Hillen H., Schroeder W., Tunggal B.;
RT   "Amino-acid sequence of human and bovine brain myelin proteolipid
RT   protein (lipophilin) is completely conserved.";
RL   Biol. Chem. Hoppe-Seyler 366:627-635(1985).
RN   [7]
RP   NUCLEOTIDE SEQUENCE.
RC   TISSUE=Spinal cord;
RX   MEDLINE=88141333; PubMed=2449536;
RA   Kronquist K.E., Crandall B.F., Macklin W.B., Campagnoni A.T.;
RT   "Expression of myelin proteins in the developing human spinal cord:
RT   cloning and sequencing of human proteolipid protein cDNA.";
RL   J. Neurosci. Res. 18:395-401(1987).
RN   [8]
RP   VARIANT PMD SER-217.
RX   MEDLINE=89371750; PubMed=2773936;
RA   Gencic S., Abuelo D., Ambler M., Hudson L.D.;
RT   "Pelizaeus-Merzbacher disease: an X-linked neurologic disorder of
RT   myelin metabolism with a novel mutation in the gene encoding
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RT   proteolipid protein.";
RL   Am. J. Hum. Genet. 45:435-442(1989).
RN   [9]
RP   VARIANT PMD LEU-15.
RX   MEDLINE=90083280; PubMed=2480601;
RA   Trofatter J., Dlouhy S.R., Demyer W., Conneally P.M., Hodes M.E.;
RT   "Pelizaeus-Merzbacher disease: tight linkage to proteolipid protein
RT   gene exon variant.";
RL   Proc. Natl. Acad. Sci. U.S.A. 86:9427-9430(1989).
RN   [10]
RP   VARIANT PMD ILE-156.
RX   MEDLINE=91214553; PubMed=1708672;
RA   Weimbs T., Dick T., Stoffel W., Boltshauser E.;
RT   "A point mutation at the X-chromosomal proteolipid protein locus in
RT   Pelizaeus-Merzbacher disease leads to disruption of myelinogenesis.";
RL   Biol. Chem. Hoppe-Seyler 371:1175-1183(1990).
RN   [11]
RP   VARIANT PMD ILE-156.
RX   MEDLINE=91189235; PubMed=1707231;
RA   Pratt V.M., Trofatter J.A., Schinzel A., Dlouhy S.R., Conneally P.M.,
RA   Hodes M.E.;
RT   "A new mutation in the proteolipid protein (PLP) gene in a German
RT   family with Pelizaeus-Merzbacher disease.";
RL   Am. J. Med. Genet. 38:136-139(1991).
RN   [12]
RP   VARIANT PMD PHE-219.
RX   MEDLINE=91352028; PubMed=1715570;
RA   Pham-Dinh D., Popot J.-L., Bosepflug-Tanguy O., Landrieu P.,
RA   Deleuze P., Boue J., Jolles P., Dautigny A.;
RT   "Pelizaeus-Merzbacher disease: a valine to phenylalanine point
RT   mutation in a putative extracellular loop of myelin proteolipid.";
RL   Proc. Natl. Acad. Sci. U.S.A. 88:7562-7566(1991).
RN   [13]
RP   VARIANTS PMD ARG-74 AND HIS-203.
RX   MEDLINE=92303562; PubMed=1376966;
RA   Doll R., Natowicz M.R., Schiffmann R., Smith F.I.;
RT   "Molecular diagnostics for myelin proteolipid protein gene mutations
RT   in Pelizaeus-Merzbacher disease.";
RL   Am. J. Hum. Genet. 51:161-169(1992).
RN   [14]
RP   VARIANTS PMD PRO-182 AND PRO-224.
RX   MEDLINE=93035344; PubMed=1384324;
RA   Strautnieks S., Rutland P., Winter R.M., Baraitser M., Malcolm S.;
RT   "Pelizaeus-Merzbacher disease: detection of mutations Thr181-->Pro and
RT   Leu223-->Pro in the proteolipid protein gene, and prenatal
RT   diagnosis.";
RL   Am. J. Hum. Genet. 51:871-878(1992).
RN   [15]
RP   VARIANT PMD GLU-166.
RX   MEDLINE=93276877; PubMed=7684886;
RA   Pratt V.M., Kiefer J.R., Lahdetie J., Schleutker J., Hodes M.E.,
RA   Dlouhy S.R.;
RT   "Linkage of a new mutation in the proteolipid protein (PLP) gene to
RT   Pelizaeus-Merzbacher disease (PMD) in a large Finnish kindred.";
RL   Am. J. Hum. Genet. 52:1053-1056(1993).
RN   [16]
RP   VARIANT PMD SER-217.
RX   MEDLINE=93176327; PubMed=7679906;
RA   Otterbach B., Stoffel W., Ramaekers V.;
RT   "A novel mutation in the proteolipid protein gene leading to
RT   Pelizaeus-Merzbacher disease.";
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RL   Biol. Chem. Hoppe-Seyler 374:75-83(1993).
RN   [17]
RP   VARIANT PMD CYS-221.
RX   MEDLINE=93258343; PubMed=7683951; DOI=10.1093/hmg/2.1.19;
RA   Iwaki A., Muramoto T., Iwaki I., Furumi H., Dario-Deleon M.L.,
RA   Tateishi J., Fukumaki Y.;
RT   "A missense mutation in the proteolipid protein gene responsible for
RT   Pelizaeus-Merzbacher disease in a Japanese family.";
RL   Hum. Mol. Genet. 2:19-22(1993).
RN   [18]
RP   VARIANT PMD SER-216.
RX   MEDLINE=94311323; PubMed=8037216;
RA   Pratt V.M., Boyadjiev S., Dlouhy S.R., Silver K., der Kaloustian V.M.,
RA   Hodes M.E.;
RT   "Comparison of statistics for candidate-gene association studies using
RT   cases and parents.";
RL   Am. J. Hum. Genet. 55:402-404(1994).
RN   [19]
RP   VARIANT SPG2 TYR-140.

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